

# COMBINATORICS OF PEDIGREES

BHALCHANDRA D. THATTE

**ABSTRACT.** A pedigree is a directed graph in which each vertex (except the founder vertices) has two parents. The main result in this paper is a construction of an infinite family of counter examples to a reconstruction problem on pedigrees, thus negatively answering a question of Steel and Hein. Some positive reconstruction results are also presented. The problem of counting distinct (mutually non-isomorphic) pedigrees is considered. The known lower and upper bounds on the number of pedigrees are improved upon, and their relevance to pedigree reconstruction from DNA sequence data is discussed. It is shown that the information theoretic bound on the number of segregating sites in the sequence data that is minimally essential for reconstructing pedigrees would not significantly change with improved enumerative estimates.

## 1. INTRODUCTION

A *general pedigree*  $\mathcal{T}(X_0)$  on a set  $X_0$ , is a finite directed graph on a vertex set  $V$  that satisfies the following conditions:

- (1) each vertex has out-degree 0 or 2;
- (2)  $X_0$  is a subset of  $V$ , and each vertex in  $X_0$  has in-degree 0;
- (3) there are no isolated vertices.

The vertices with out-degree 0 are called the *founders*. The vertices in  $X_0$  are called *extant*. The cardinality of  $X_0$  is called the *order* of the pedigree. Note that  $X_0$  is a subset of the set of vertices with in-degree 0.

A *discrete generation pedigree* on  $X_0$  is a pedigree on vertex set  $V = \cup_{i=0}^d X_i$ , where  $X_i$  are disjoint sets,  $X_d$  is the set of founders, and every vertex  $u$  in  $X_i; i < d$  has outgoing arcs  $uv$  and  $uw$  to vertices  $v$  and  $w$ , respectively, in  $X_{i+1}$ . In this case,  $d$  is the *depth* of the pedigree.

If there is an arc from a vertex  $u$  to a vertex  $v$ , then  $v$  is called a parent of  $u$ , and  $u$  is called a child of  $v$ . If there is a directed path from a vertex  $u$  to a vertex  $v$  in a pedigree, then  $v$  is said to be an *ancestor*

---

*Date:* 10 September, 2006.

*2000 Mathematics Subject Classification.* Primary: 05C60.

*Key words and phrases.* pedigrees, reconstruction, enumeration.

of  $u$ , and  $u$  is said to be a *descendent* of  $v$ . Trivially, each vertex is its own ancestor as well as its own descendent, but not its own parent or child. If there is a directed path  $u - u_1 - \dots - u_k$  then  $u_k$  is called a  $k$ -th grandparent of  $u$ , and  $u$  is called a  $k$ -th grandchild of  $u_k$ .

A pedigree  $\mathcal{P}(X_0)$  with vertex set  $U$  is said to be *isomorphic* to a pedigree  $\mathcal{Q}(Y_0)$  with vertex set  $V$  if there is a one-one map  $f : U \rightarrow V$  such that  $u_1 - u_2$  is an arc in  $\mathcal{P}(X_0)$  if and only if  $f(u_1) - f(u_2)$  is an arc in  $\mathcal{Q}(Y_0)$ . Although this is a standard definition of graph isomorphism, we will be interested in pedigrees in which the extant vertices are labelled. Therefore, if  $X_0 = Y_0 = \{x_i; 1 \leq i \leq n\}$  then we will be interested only in isomorphisms  $\pi$  for which  $\pi(x_i) = x_i$  for all  $1 \leq i \leq n$ .

A motivation to study pedigrees comes from biology, where one is interested in reconstructing pedigrees of populations. But it is hoped that the main result in this paper - the non-reconstructibility of pedigrees from sub-pedigrees - will also be of interest to combinatorialists interested in the well known reconstruction conjectures.

Steel and Hein [3] posed and partially solved reconstruction and enumeration questions about pedigrees. Motivated by results in phylogenetics, a natural question to ask is: is a pedigree determined up to isomorphism from the pairwise distances between extant vertices? A pair of extant vertices  $x$  and  $y$  in a pedigree may have several common ancestors, therefore, it is assumed that all possible distances (in the undirected sense) between all pairs of extant vertices are given. Such a question is not expected to have a positive answer, as demonstrated by a counter example in [3]. Despite the counter example, variations of this question are definitely significant in evolutionary biology. Steel and Hein considered the following weaker question.

Let  $\mathcal{P}(X_0)$  be a pedigree. A *sub-pedigree*  $\mathcal{P}(Y)$  of  $\mathcal{P}(X_0)$  is obtained by deleting every vertex in  $\mathcal{P}(X_0)$  that has no descendent in  $Y$ . Now if sub-pedigrees on all two-element subsets of  $X_0$  are given up to isomorphism, can we construct the sub-pedigree on  $X_0$  up to isomorphism? Steel and Hein presented a counter example in their paper. They posed the following problem.

**Problem 1.** Is there an integer  $r > 2$  such that every pedigree  $\mathcal{P}(X_0)$  of order  $n > r$  determined up to isomorphism if all its sub-pedigrees  $\mathcal{P}(Y)$  such that  $|Y| = r$  are given up to isomorphism?

Combinatorialists familiar with the reconstruction conjectures might be tempted to dismiss this question, therefore, it must be pointed out that the set  $X_0$  in a pedigree is labelled. In other words, “a sub-pedigree  $\mathcal{P}(Y)$  given up to isomorphism” is to be interpreted as a pedigree in

which all vertices except the ones in  $Y$  are unlabelled. The following definitions are introduced to make this remark more formal.

**Definition 1.** Let  $n > r > 2$  be positive integers. Let  $\mathcal{T}(X_0)$  and  $\mathcal{U}(X_0)$  be two pedigrees of order  $n$ . The two pedigrees are said to be *r-hypomorphic* to each other if for every  $Y \subset X_0; |Y| = r$ , there is an isomorphism  $\pi_Y$  from the sub-pedigree  $\mathcal{T}(Y)$  of  $\mathcal{T}(X_0)$  to the sub-pedigree  $\mathcal{U}(Y)$  of  $\mathcal{U}(X_0)$  such that  $\pi_Y(x) = x$  for all  $x \in Y$ . A pedigree  $\mathcal{T}(X_0)$  is said to be *r-reconstructible* if for every pedigree  $\mathcal{U}(X_0)$  that is *r-hypomorphic* to  $\mathcal{T}(X_0)$ , there is an isomorphism  $\pi$  from  $\mathcal{T}(X_0)$  to  $\mathcal{U}(X_0)$  such that  $\pi(x) = x$  for all  $x \in X_0$ .

**Problem 2.** Is there an integer  $r > 2$  such that all pedigrees of order  $n > r$  are *r-reconstructible*?

In Section 2, we present a family of counter examples as well as a few positive results on constant population size pedigrees. We prove that for every  $n > 3$ , there are pedigrees of order  $n$  that are not even  $(n-1)$ -reconstructible. The problem of classification of non-reconstructible pedigrees remains open, and we suspect that it might have an algebraic structure similar to the Nash-Williams' lemma in edge reconstruction theory, see [2].

Steel and Hein considered the question of enumerating mutually non-isomorphic pedigrees of a fixed depth. A lower bound on the number of distinct pedigrees implies, by an information theoretic argument, a lower bound on the number of segregating DNA sites that would be necessary in order to reconstruct the pedigree of a population from the sequence data. In Section 3 we prove tighter lower and upper bounds, and show that the information theoretic lower bound does not increase much. Steel and Hein leave the problem of enumerating general pedigrees open. Here we enumerate general pedigrees as well, and again show that purely information theoretic arguments as in their paper are not sufficient to show that general pedigrees would necessarily require significantly more segregating sites for their reconstruction from the sequence data.

## 2. RECONSTRUCTION OF PEDIGREES.

**2.1. A negative result.** We solve Problem 1 negatively by constructing an infinite family of pairs of non-isomorphic pedigrees that have correspondingly isomorphic sub-pedigrees. That is, we prove the following

**Theorem 1.** *For every  $n > 2$ , there are non-isomorphic pedigrees  $\mathcal{T}(X_0)$  and  $\mathcal{U}(X_0)$  of order  $n$  that are  $(n-1)$ -hypomorphic.*

*Proof.* The proof is divided in two cases. The case  $n = 3$  gives the basic idea, which is then generalised to arbitrary values of  $n$ .

**Case  $n = 3$ .**

Consider the non-isomorphic graphs  $K_{1,3}$  and  $K_3$ . Let the edges of both graphs be arbitrarily labelled  $e_1, e_2, e_3$ , where, following the standard graph theoretic convention, an edge is a set of two vertices. It is clear that  $K_{1,3} - e_i \cong K_3 - e_i$  for all  $i$ , where  $-e_i$  denotes deletion of the edge  $e_i$  and the resulting isolated vertices. Now suppose that the end vertices of each edge  $e_i$  are parents of the vertex  $x_i \in X_0$  in each of the pedigrees  $\mathcal{T}(X_0)$  and  $\mathcal{U}(X_0)$ . Then the pedigrees  $\mathcal{T}(\{x_i, x_j\})$  and  $\mathcal{U}(\{x_i, x_j\})$  are isomorphic for all  $i, j$ , but the pedigrees  $\mathcal{T}(X_0)$  and  $\mathcal{U}(X_0)$  are not isomorphic. This example proves the theorem for  $n = 3$ . The pedigrees  $\mathcal{T}(X_0), \mathcal{U}(X_0)$ , and their sub-pedigrees are shown in Figure 1.

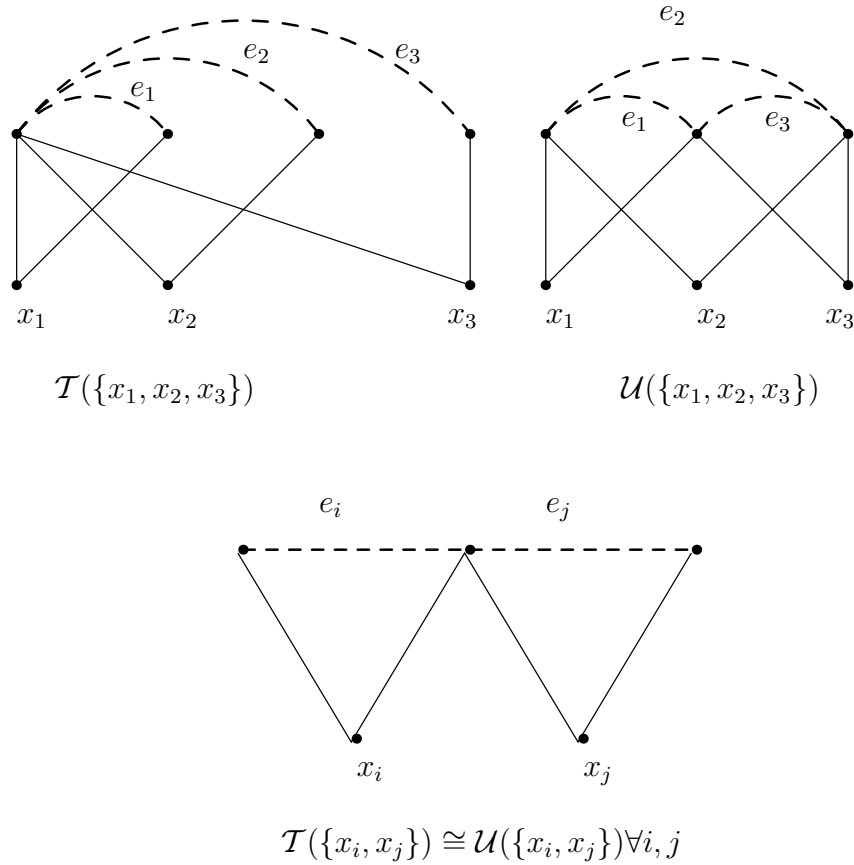


FIGURE 1. Pedigrees based on  $K_{1,3}$  and  $K_3$

**Case  $n > 3$** 

We have to construct hypergraphs that play the role that  $K_{1,3}$  and  $K_3$  play above. We construct a hypergraph  $G$  with edge set  $\{g_i; 1 \leq i \leq n\}$  and a hypergraph  $H$  with edge set  $\{h_i; 1 \leq i \leq n\}$  such that the following conditions are satisfied.

- (1)  $G \not\cong H$
- (2) For each  $i; 1 \leq i \leq n$ ,  $G - g_i \cong H - h_i$ ; moreover, there is an isomorphism between  $G - g_i$  and  $H - h_i$  that *preserves the edge order*, that is, vertices in an edge  $g_j$  in  $G - g_i$  are mapped to vertices in  $h_j$  under such an isomorphism, for each  $j \neq i$ .

Once such hypergraphs are constructed, we treat each edge in each hypergraph as a founder set. We construct pedigrees  $T_i$  on founder sets  $g_i$ , and pedigrees  $U_i$  on founder sets  $h_i$  such that

- (1) pedigrees  $T_i; 1 \leq i \leq n$  are vertex-disjoint except possibly for their founder sets  $g_i$ ;
- (2) pedigrees  $U_i; 1 \leq i \leq n$  are vertex-disjoint except possibly for their founder sets  $h_i$ ;
- (3) pedigrees  $T_i$  and  $U_i$  are correspondingly isomorphic; moreover, for all  $i, j; i \neq j$ , an isomorphism between  $G - g_j$  and  $H - h_j$  that preserves the edge order extends to an isomorphism between  $T_i$  and  $U_i$ ;
- (4) each of the pedigrees  $T_i$  and  $U_i$  contains exactly one extant vertex  $x_i$ .

The resulting pedigrees  $\cup_{i=1}^n T_i$  and  $\cup_{i=1}^n U_i$  are non-isomorphic (since the hypergraphs  $G$  and  $H$  are non-isomorphic) but their sub-pedigrees are correspondingly isomorphic.

**Construction of hypergraphs  $G$  and  $H$** 

The required hypergraphs are constructed by a simple application of linear algebra.

Let each integer in  $\{0, 2^n - 1\}$  be written in base 2 as an  $n$ -digit number by padding sufficiently many zeros on the left. We count its digits from the right. The set of  $n$ -digit binary numbers is denoted by  $[2^n]$ . The  $i$ 'th digit of a number  $k$  is denoted by  $k(i)$ , and the number obtained by setting the  $i$ 'th digit of  $k$  to 0 (or 1) is denoted by  $k(i \leftarrow 0)$  (or, respectively,  $k(i \leftarrow 1)$ ). The number of ones and the number of zeros in  $k$  are denoted by  $\#1(k)$  and  $\#0(k)$ , respectively.

The isomorphism class of a hypergraph  $G$  with edge set  $\{g_i; 1 \leq i \leq n\}$  may be represented by a list of integers  $a(k); k \in [2^n]$ , where  $a(k)$  is the number of vertices in  $\cap_{i=1}^n f_i$ , where  $f_i = g_i$  if  $k(i) = 1$ , and  $f_i = \bar{g}_i$ , (that is, the complement of  $g_i$ ), if  $k(i) = 0$ . In other words, we have to only specify the number of vertices in each region of the Venn diagram

of  $g_i$ ;  $1 \leq i \leq n$ . Let the list of integers  $b(k); k \in [2^n]$  similarly denote the isomorphism class of  $H$ .

The condition  $G - g_i \cong H - h_i$  for  $1 \leq i \leq n$ , with an isomorphism between them that preserves the edge order, may be expressed as

$$(1) \quad a(k(i \leftarrow 0)) + a(k(i \leftarrow 1)) = b(k(i \leftarrow 0)) + b(k(i \leftarrow 1)); k \in [2^n]$$

Since we are interested in non-isomorphic hypergraphs  $G$  and  $H$ , we must find solutions to the above equations so that  $a(k) \neq b(k)$  for some  $k \in [2^n]$ .

We verify that

$$(2) \quad \begin{array}{lll} a(k) = 1, & b(k) = 0 & \text{when } k \text{ has even number of 1's} \\ a(k) = 0, & b(k) = 1 & \text{when } k \text{ has odd number of 1's} \end{array}$$

satisfy Equations (1).

It can be easily verified that  $K_{1,3}$  and  $K_3 \cup K_1$  do in fact satisfy the above solutions, where we include an isolated vertex in one of the graphs purely for algebraic convenience.

Now on we write  $[2^n] = [2^n]_e \cup [2^n]_o$ , where  $[2^n]_e$  is the set of integers having an even number of 1's in their binary representation, and  $[2^n]_o$  is the set of integers having an odd number of 1's in their binary representation. In this notation, the hypergraphs  $G$  and  $H$  are described as follows: the set  $[2^n]_e$  is the set of vertices of  $G$ , and a vertex  $k \in [2^n]_e$  is in  $g_i$  if and only if  $k(i) = 1$ . Similarly, the set  $[2^n]_o$  is the vertex set of  $H$ , and a vertex  $k \in [2^n]_o$  is in edge  $h_i$  if and only if  $k(i) = 1$ .

The vertex  $k = 0$  is in  $G$ , but is an isolated vertex, and is included at this stage only for algebraic convenience, and may be deleted after completing the construction of non-reconstructible pedigrees.

It is clear that  $G$  and  $H$  are non-isomorphic, since each of them has  $2^{n-1}$  vertices, but  $G$  has the isolated vertex 0, while  $H$  has no isolated vertex. What is an isomorphism between  $G - g_i$  and  $H - h_i$ ? An edge order preserving isomorphism from  $G - g_i$  to  $H - h_i$  must map vertices in a region of the Venn diagram of  $\cup_{j|j \neq i} g_j$  to the corresponding region of the Venn diagram of  $\cup_{j|j \neq i} h_j$ . Consider any  $k \in [2^n]$ . The vertex  $k(i \leftarrow 0)$  is in  $g_j$  for some  $j \neq i$  if and only if the vertex  $k(i \leftarrow 1)$  is in  $h_j$ , because the two vertices differ only in their  $i$ 'th digit. Therefore, if  $k(i \leftarrow 0)$  is in  $G$ , then an edge order preserving isomorphism between  $G - g_i$  and  $H - h_i$  must map the vertex  $k(i \leftarrow 0)$  to the vertex  $k(i \leftarrow 1)$ . Similarly, if the vertex  $k(i \leftarrow 1)$  is in  $G$ , then an edge order preserving isomorphism between  $G - g_i$  and  $H - h_i$  must map the vertex  $k(i \leftarrow 1)$  to the vertex  $k(i \leftarrow 0)$ . Moreover, this isomorphism is unique. On the standard hypercube on  $[2^n]$ , each vertex in  $G - g_i$  is mapped to its neighbour along the  $i$ 'th axis, which is in  $H - h_i$ .

**Example 1.** Let  $n = 4$ , and let the hypergraphs  $G$  and  $H$  be defined on vertex sets  $[2^4]_e$  and  $[2^4]_o$  as follows:

$$\begin{aligned} g_1 &= \{0011, 0101, 1001, 1111\}, g_2 = \{0011, 0110, 1010, 1111\}, \\ g_3 &= \{0101, 0110, 1100, 1111\}, g_4 = \{1001, 1010, 1100, 1111\}, \\ h_1 &= \{0001, 0111, 1011, 1101\}, h_2 = \{0010, 0111, 1011, 1110\}, \\ h_3 &= \{0100, 0111, 1101, 1110\}, h_4 = \{1000, 1011, 1101, 1110\}, \end{aligned}$$

where  $g_i$  are the edges of  $G$  and  $h_i$  are the edges of  $H$ . The isomorphism  $\pi_1$  from  $G - g_1$  to  $H - h_1$  that preserves the edge order is given by  $\pi_1(0000) = 0001$ ,  $\pi_1(0011) = 0010$ ,  $\pi_1(0101) = 0100$ ,  $\pi_1(1001) = 1000$ ,  $\pi_1(0110) = 0111$ ,  $\pi_1(1010) = 1011$ ,  $\pi_1(1100) = 1101$ ,  $\pi_1(1111) = 1110$ . Observe that  $\pi_1(g_2) = h_2$ ,  $\pi_1(g_3) = h_3$ , and  $\pi_1(g_4) = h_4$  under this map.

### Construction of $T_i$ and $U_i$

As stated earlier, for each  $i$ , pedigrees  $T_i$  and  $U_i$  must be so constructed that (the unique) edge order preserving isomorphism between  $G - g_j$  and  $H - h_j$  extends to an isomorphism between  $T_i$  and  $U_i$  for all  $j \neq i$ .

Let a balanced binary tree  $T_i$  be defined so that  $x_i$  is its root and  $g_i$  is its set of leaves. By convention, the root  $x_i$  is the lowest vertex (at depth 0) in  $T_i$ , and the leaves are the highest vertices (at depth  $n - 2$ ) in  $T_i$ . For a vertex  $t$  in  $T_i$ , let  $T_i(t)$  be the subtree of  $T_i$  induced by  $t$  and all vertices in  $T_i$  that are above  $t$ . Let  $t_0$  and  $t_1$  be the parents of  $t$ . The subtree  $T_i(t)$  is a union of subtrees  $L(t)$  and  $R(t)$ , where  $L(t)$  is induced by vertices  $t$ ,  $t_0$ , and all vertices above  $t_0$ , and the subtree  $R(t)$  is induced by vertices  $t$ ,  $t_1$ , and all vertices above  $t_1$ . We call  $L(t)$  the *left subtree* at  $t$ , and  $R(t)$  the *right subtree* at  $t$ .

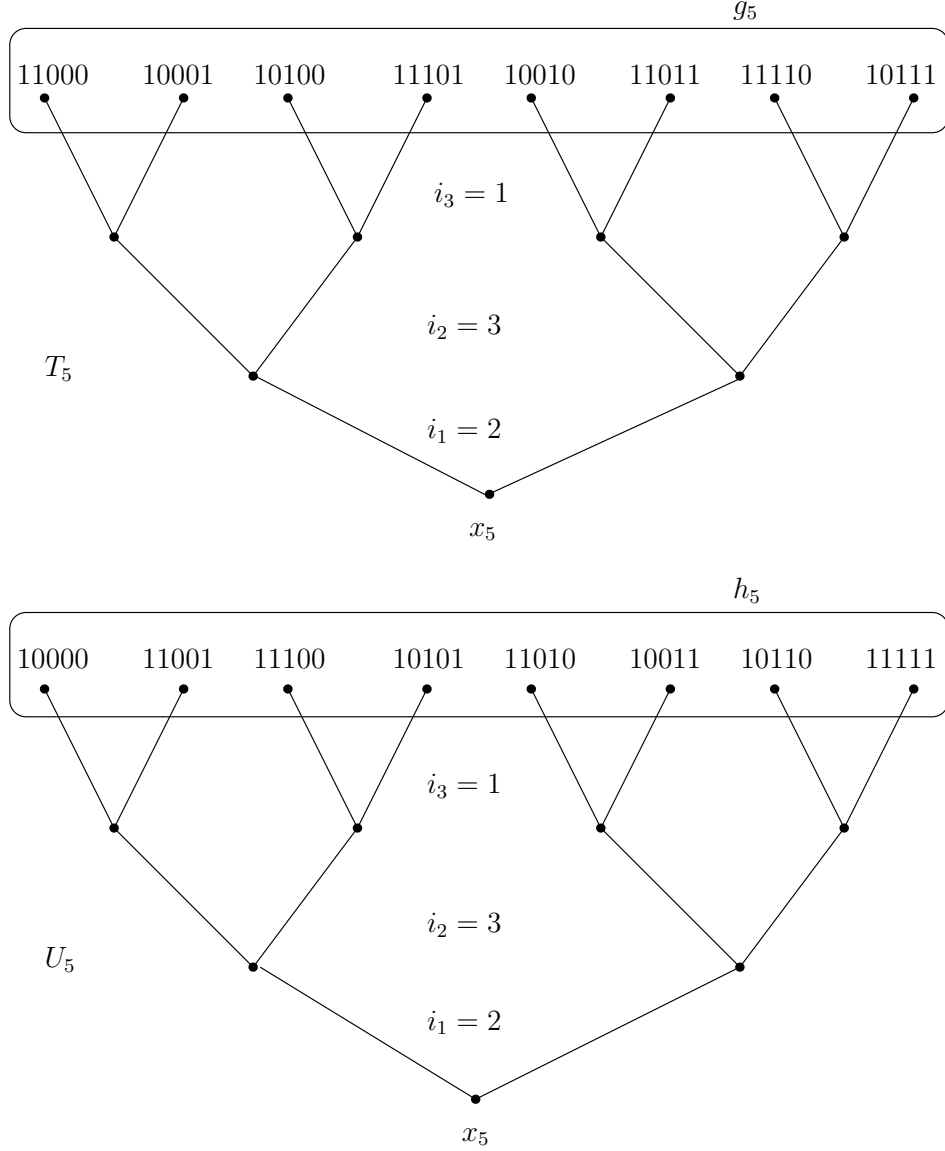
Let  $i_1, i_2, \dots, i_{n-1}$  be the integers  $1 \leq j \leq n; j \neq i$  in arbitrary order. The vertices in  $g_i$  are grouped in such a way that for each vertex  $t$  at depth  $k; 0 \leq k \leq n - 3$ , if a vertex  $p \in g_i$  is a leaf of  $L(t)$  then  $p(i_{k+1}) = 0$ , and if a vertex  $p \in g_i$  is a leaf of  $R(t)$  then  $p(i_{k+1}) = 1$ .

The vertices in  $h_i$  are partitioned, and a binary tree  $U_i$  is constructed analogously for the same ordering  $i_j; 1 \leq j \leq n - 1$ .

For  $n = 5$  and  $i = 5$  and the ordering  $i_1 = 2, i_2 = 3, i_3 = 1, i_4 = 4$ , the trees  $T_5$  and  $U_5$  are shown in Figure 2.

We show that for every  $j \neq i$ , the unique isomorphism between  $G - g_j$  and  $H - h_j$  extends to an isomorphism between  $T_i$  and  $U_i$ .

Let  $\bar{b} = (b_1, \dots, b_j); 0 \leq j \leq n - 2$  be a  $j$ -tuple of 0's and 1's. Extending a notation introduced earlier, let  $g_i(\bar{b})$  denote the set  $\{k \in g_i | k(i_1) = b_1, k(i_2) = b_2, \dots, k(i_j) = b_j\}$ , which is the set of leaves of a binary subtree of  $T_i$  rooted at the vertex  $t(\bar{b})$  at depth  $j$ . For example,

FIGURE 2. Binary pedigrees  $T_5$  and  $U_5$  for  $n = 5$ 

when  $\bar{b} = (0)$ ,  $g_i(\bar{b})$  is the set of leaves above the left parent of  $x_i$ , and  $t(\bar{b})$  is the left parent of  $x_i$ . The set  $h_i(\bar{b})$  and the vertex  $u(\bar{b})$  are analogously defined for  $U_i$ . By convention, an empty tuple  $\bar{b}$  defines the sets  $g_i$  and  $h_i$ , and the trees  $T_i$  and  $U_i$ , rooted at  $x_i$ ; and a tuple  $\bar{b}$  of length  $n - 2$  defines singleton subsets  $\{t(\bar{b})\}$  of  $g_i$ , and  $\{u(\bar{b})\}$  of  $h_i$ .



A tuple  $\bar{b}$  of length  $n - 2$  also uniquely determines the digits  $t(\bar{b})(i_{n-1})$  and  $u(\bar{b})(i_{n-1})$ , since we know that  $\#1(t(\bar{b}))$  is even and  $\#1(u(\bar{b}))$  is odd. Also, if  $t(\bar{b}(i_{n-1})) = 1$  then  $u(\bar{b}(i_{n-1})) = 0$ , and if  $t(\bar{b}(i_{n-1})) = 0$  then  $u(\bar{b}(i_{n-1})) = 1$ . Therefore, the map  $t(\bar{b}) \longrightarrow u(\bar{b})$  for all tuples of length at most  $n - 2$  extends the isomorphism between  $G - g_{i_{n-1}}$  and  $H - h_{i_{n-1}}$ .

Let  $\bar{b}$  be a tuple as above. Extending the notation  $k(i \leftarrow 0)$  to  $\bar{b}$ , we define  $\bar{b}(i \leftarrow 0)$  to be the tuple obtained by setting  $b_i = 0$  in  $\bar{b}$ , and  $\bar{b}(i \leftarrow 1)$  to be the tuple obtained by setting  $b_i = 1$  in  $\bar{b}$ .

Let  $\bar{b}$  be a tuple of length  $n - 2$  and  $j \leq n - 2$ . By an argument as in the above paragraph, we have

- (1) if  $t(\bar{b}(j \leftarrow 0)(i_{n-1})) = 1$  then  $u(\bar{b}(j \leftarrow 1)(i_{n-1})) = 1$ ;
- (2) if  $t(\bar{b}(j \leftarrow 0)(i_{n-1})) = 0$  then  $u(\bar{b}(j \leftarrow 1)(i_{n-1})) = 0$ ;
- (3) if  $t(\bar{b}(j \leftarrow 1)(i_{n-1})) = 1$  then  $u(\bar{b}(j \leftarrow 0)(i_{n-1})) = 1$ ;
- (4) if  $t(\bar{b}(j \leftarrow 1)(i_{n-1})) = 0$  then  $u(\bar{b}(j \leftarrow 0)(i_{n-1})) = 0$ .

Therefore, for each  $j$ ;  $1 \leq j \leq n - 2$ , the map defined by

- (1)  $t(\bar{b}) \longrightarrow u(\bar{b})$  for all tuples of length at most  $j - 1$ ;
- (2)  $t(\bar{b}(j \leftarrow 0)) \longrightarrow u(\bar{b}(j \leftarrow 1))$  for all tuples of length at least  $j$ ;
- and
- (3)  $t(\bar{b}(j \leftarrow 1)) \longrightarrow u(\bar{b}(j \leftarrow 0))$  for all tuples of length at least  $j$

extends the isomorphism between  $G - g_j$  and  $H - h_j$ . Observe that this map sends the vertices in the left subtree  $L(t(\bar{b}))$  in  $T_i$  to the vertices in the right subtree  $R(u(\bar{b}))$  in  $U_i$ , and the vertices in the right subtree  $R(t(\bar{b}))$  in  $T_i$  to the vertices in the left subtree  $L(u(\bar{b}))$  in  $U_i$ , for each tuple  $\bar{b}$  of length  $j$ . Since the trees  $T_i$  and  $T_j$  (and trees  $U_i$  and  $U_j$ ) are disjoint except for their founders for all  $i \neq j$ , the isomorphism between  $G - g_j$  and  $H - h_j$  extends to an isomorphism between pedigrees  $\mathcal{T}(X_0 \setminus \{x_j\})$  and  $\mathcal{U}(X_0 \setminus \{x_j\})$  for all  $j$ .

An isomorphism between  $G - g_{i_2}$  and  $H - h_{i_2}$  that extends to an isomorphism between  $T_i$  and  $U_i$ ;  $i \neq i_2$  is schematically shown in Figure 3.  $\square$

*Remark 1.* The pedigrees constructed above do not admit a valid gender labelling. That is, we cannot assign labels  $m$  (male) and  $f$  (female) to all vertices so that each vertex (except founders) has one male parent and one female parent. For example, in the  $n = 3$  case,  $K_3$  is not a bipartite graph, so a valid gender labelling is impossible. But the examples can be easily modified to create non-reconstructible pedigrees that also admit valid gender labels. Each vertex in a pedigree constructed above may be duplicated, and one vertex may be treated male and the other female, as shown in Figure 4. At the bottom of the tree  $T_i$  (or

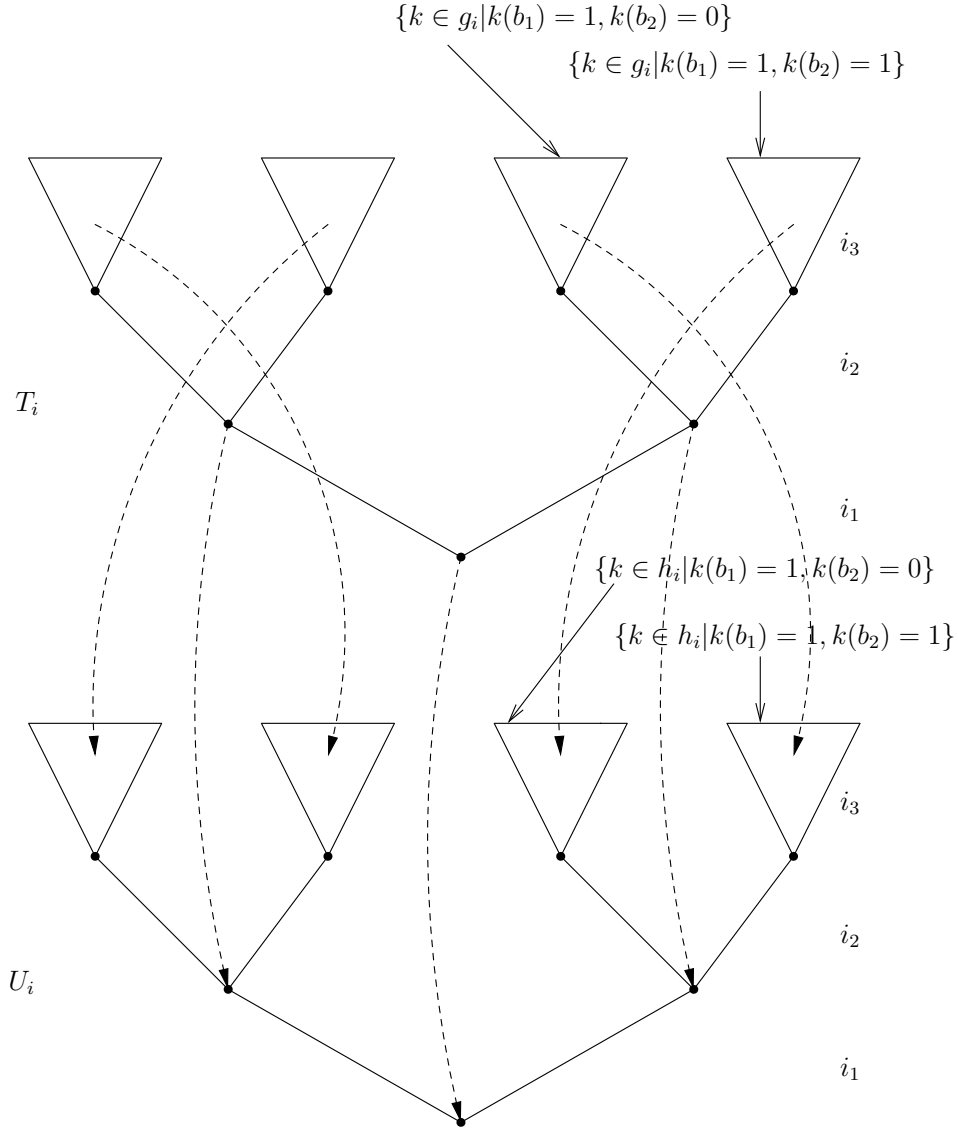


FIGURE 3. Isomorphism between  $G - g_{i_2}$  and  $H - h_{i_2}$  extends to an isomorphism between  $T_i$  and  $U_i$

$U_i$ ), the vertex  $x_i$  is duplicated as vertices  $x_i^m$  and  $x_i^f$ , and the new vertex  $x_i$  is a child of  $x_i^m$  and  $x_i^f$ .

*Remark 2.* Let  $k, k' \in [2^n]$  be any two adjacent vertices on the hypercube. From Equation (1), if  $a(k) - b(k) = p > 0$  for some  $p$ , then

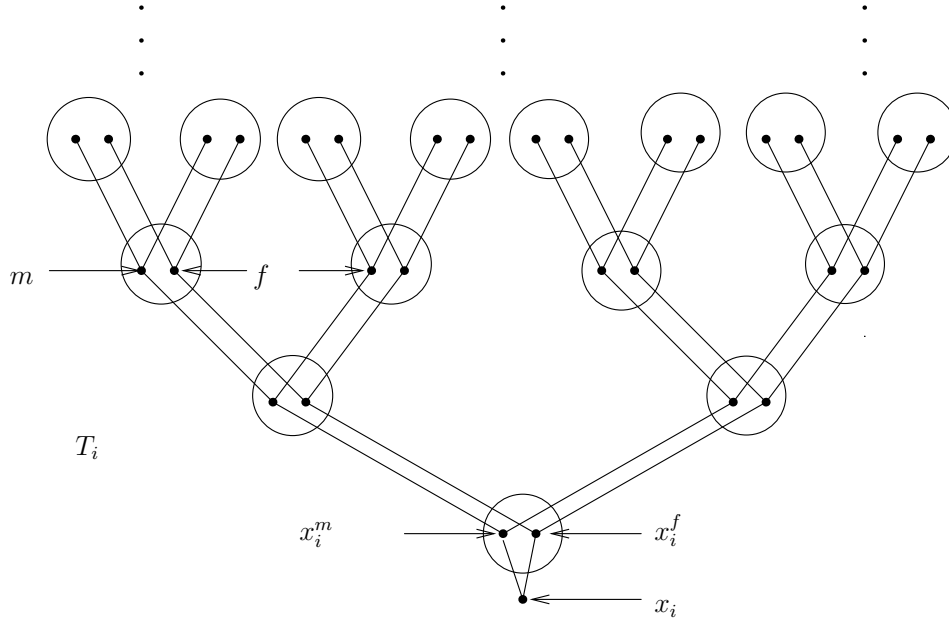


FIGURE 4. Construction of a pedigree with a valid gender labelling

$b(k') - a(k') = p$ , regardless of which digit  $k$  and  $k'$  differ at. In fact, by connectivity of the hypercube, we have  $a(r) - b(r) = p$  for all vertices  $r \in [2^n]$  that are at even distance from  $k$  on the hypercube, and  $b(r) - a(r) = p$  for all vertices  $r \in [2^n]$  that are at odd distance from  $k$  on the hypercube. This further implies that the hypergraphs  $G$  and  $H$  constructed in the above counter example have a special structure: for each  $i$ ;  $1 \leq i \leq n$ ,  $|g_i| = |h_i| \geq 2^{n-2}$ . Let  $G(d)$  be the hypergraph with edge set  $\{g_i(d); 1 \leq i \leq n\}$ , where  $g_i(d)$  is the set of grandparents of  $x_i$  at depth  $d$  in the pedigree  $\mathcal{T}(X_0)$ , and let  $H(d)$  and  $h_i(d)$  be similarly defined for the pedigree  $\mathcal{U}(X_0)$ , then the hypergraphs  $G(d)$  and  $H(d)$  must be isomorphic whenever  $d < n - 2$ .

We end this subsection with a conjecture motivated by the observations made in Remark 2.

**Conjecture 1.** *The counter example constructed in Theorem 1 is minimal. In other words, if a pedigree  $\mathcal{T}(X_0)$  of order  $n$  is not  $(n - 1)$ -reconstructible then it has depth at least  $n - 2$ , and there are at least  $2^{n-1}$  ancestors at depth  $n - 2$ .*

*Remark 3.* Let  $G$  and  $H$  be simple graphs with edge sets  $E(G) = \{g_i; 1 \leq i \leq m\}$  and  $E(H) = \{h_i; 1 \leq i \leq m\}$ , respectively, such that  $G - g_i \cong H - h_i$  for all  $1 \leq i \leq m$ . Then the edge reconstruction conjecture states that  $G \cong H$  provided  $m > 3$ . The condition  $m > 3$  is required since  $K_{1,3}$  and  $K_3$  - the graphs used as the base case of our construction of non-reconstructible pedigrees - are not *edge reconstructible*. Although no counter examples are yet known, Nash-Williams [2] proved a characterisation of (hypothetical) counter examples to edge reconstruction. His characterisation was based on a generalisation of ideas earlier introduced by Lovász [1]. Without going into details, we note that the counter examples presented here have certain similarities with the characterisation by Nash-Williams. It may be possible to exploit such similarities to prove Conjecture 1.

**2.2. A positive result.** Let  $\mathcal{T}(X_0)$  be a discrete generation pedigree on  $X_0$  of order  $n > 2$ . Let  $S_{n-1}(\mathcal{T}) = \{\mathcal{T}(Y) | Y \subset X_0, |Y| = n - 1\}$ . Consider the edge labelled (multi) graph  $G_1$  whose vertex set is  $X_1$  (that is, the vertices at depth 1), and vertices  $x, y \in X_1$  are joined by an edge  $e_i$  if they are the parents of  $x_i$ .

**Lemma 1.** *If there are vertices  $x_i$  and  $x_j$  in  $X_0$  that have the same parents, then  $\mathcal{T}(X_0)$  is uniquely determined by  $S_{n-1}(\mathcal{T})$ .*

*Proof.* The situation in the lemma is recognised by looking at  $\mathcal{T}(X_0 \setminus x_k)$ , where  $x_k \notin \{x_i, x_j\}$ . Now  $\mathcal{T}(X_0)$  is uniquely obtained from  $\mathcal{T}(X_0 \setminus x_i)$  by joining  $x_i$  to the parents of  $x_j$ .  $\square$

**Lemma 2.** *If  $n > 3$  and if  $G_1$  contains a cycle then  $\mathcal{T}(X_0)$  is uniquely determined  $S_{n-1}(\mathcal{T})$ .*

*Proof.* Let  $e_i$  be an edge in a cycle in  $G_1$ . The end vertices of  $e_i$  are the two parents of  $x_i$ . Since the set of half brothers of  $x_i$  is known from the collection  $S_{n-1}$ , the parents of  $x_i$  are uniquely recognised in  $\mathcal{T}(X \setminus x_i)$ . Note that we need the condition  $n > 3$  because otherwise we would get a counter example based on  $G_1 \cong K_3$  or  $G_1 \cong K_{1,3}$ .  $\square$

**Corollary 1.** *If  $|X_1| \leq n$  and  $n > 3$  then  $S_{n-1}(\mathcal{T})$  determines  $\mathcal{T}(X_0)$  up to congruence.*

*Proof.* If no two vertices in  $X_0$  have the same two parents then  $G_1$  has  $n$  simple edges (that is no two edges are parallel edges), and there is a cycle in  $G_1$ .  $\square$

We end this section with another conjecture.

**Conjecture 2.** *Discrete generation pedigrees of order  $n$  that have a constant population in each generation are  $r$ -reconstructible for  $r > \log n$ .*

This conjecture is true if Conjecture 1 is true. For suppose that Conjecture 1 is true but Conjecture 2 is not true, and that there is a pedigree of order  $n$  that is not  $r$ -reconstructible for some  $r > \log n$ . Therefore, for some  $r > \log n$ , there is a sub-pedigree of order  $r+1$  that is not  $r$ -reconstructible. Such a sub-pedigree must have depth at least  $r-1$ , and must have at least  $2^r$  vertices at depth  $r-1$ , implying that  $r \leq \log n$ . Thus if  $r > \log n$  then we have a contradiction, therefore, all sub-pedigrees of order  $r+1$  are  $r$ -reconstructible when  $r > \log n$ , and we can complete the reconstruction inductively.

### 3. ENUMERATION OF PEDIGREES

Let  $N(n, d)$  be the number of distinct (mutually non-isomorphic) discrete generation pedigrees of depth  $d$  with  $n$  vertices in each generation. As before, the extant vertices are assumed to be labelled, and other vertices are assumed to be unlabelled.

In a general pedigree, the depth of a vertex  $u$  is the largest integer  $k$  for which  $u$  is a  $k$ 'th grandparent of an extant vertex. The depth of a pedigree is the largest integer  $d$  for which there is a vertex of depth  $d$  in the pedigree. Let the number of distinct general pedigrees of depth  $d$  with constant number  $n$  of vertices at each depth be  $M(n, d)$ .

The purpose of this section is to derive lower and upper bounds on  $N(n, d)$  and  $M(n, d)$ . The bounds are relevant to an information theoretic argument that was used by Steel and Hein in the context of a reconstruction question.

**Theorem 2.**

$$(3) \quad \left( \frac{(n-1)n^{n-2}}{2} \right)^d \leq N(n, d) \leq \binom{n}{2}^{nd}$$

$$(4) \quad \frac{(n-1)n^{n-2}}{2} \prod_{k=0}^{d-2} ((n/2)(d-1-k))^n \leq M(n, d) \leq \binom{nd-1}{2}^{nd}$$

*Proof.* Let  $\mathcal{P}(X_0)$  be a discrete generation pedigree of depth  $d$  on  $X_0$ . Let  $X_i$  be the set of vertices at depth  $i$ . Let  $|X_i| = n$  for all  $i$ ;  $0 \leq i \leq d$ . For each  $i$ ;  $1 \leq i \leq d$ , define a graph  $G_i$  as follows: the vertex set of  $G_i$  is  $X_i$ , and  $\{u, v\}$  is an edge in  $G_i$  if  $u$  and  $v$  have a child in  $X_{i-1}$ . Thus  $1 \leq e(G_i) \leq n$  for  $1 \leq i \leq d$ , where  $e(G)$  denotes the number of edges of a graph  $G$ . We restrict ourselves to bipartite graphs  $G_i$  so that it is possible to assign valid gender labels to the vertices of pedigrees.

Let  $S(n, k)$  denote the Sterling number of the second kind. There are  $S(n, k)$  partitions of  $X_0$  in groups of siblings, where *siblings* are vertices that share both parents. If the vertices of  $G_1$  are labelled then there are  $k!$  ways of assigning the groups of siblings to pairs of parents. Therefore, each labelled graph  $G_1$  gives  $S(n, k)k!$  labelled pedigrees of depth 1. Some of those pedigrees may be isomorphic to each other since there may be automorphisms of  $G_1$  that permute the edges of  $G_1$  non-trivially. Therefore, the number of distinct pedigrees of depth 1 that can be obtained from a labelled graph  $G_1$  is given by

$$N(n, d, G_1) = \frac{S(n, k)k!}{|\text{aut}LG_1|},$$

where  $LG_1$  denotes the line graph of  $G_1$ , and  $\text{aut}G$  denotes the automorphism group of a graph  $G$ . If every non-trivial automorphism of  $G_1$  permutes the edges of  $G_1$  non-trivially then the number of distinct pedigrees of depth 1 that can be obtained from  $G_1$  is given by

$$N(n, d, G_1) = \frac{S(n, k)k!}{|\text{aut}G_1|}.$$

Each non-trivial automorphism of a graph  $G$  permutes the edges of  $G$  non-trivially if and only if  $G$  has no isolated edges and not more than one isolated vertices. Therefore,

$$N(n, 1) \geq \sum_G \frac{S(n, e(G))e(G)!}{|\text{aut}G|},$$

where the summation is over all distinct bipartite graphs  $G$  having  $n$  vertices, at least 1 and at most  $n$  edges, at most one isolated vertex, and no isolated edges.

Pedigrees of depth 1 considered above have the additional property that they have no non-trivial automorphisms that fix each vertex in  $X_0$ , implying that the vertices of  $X_1$  are distinguishable in such pedigrees. Therefore,

$$N(n, d) \geq \left( \sum_G \frac{S(n, e(G))e(G)!}{|\text{aut}G|} \right)^d,$$

where the summation is over all graphs of the type described above.

Summing over only graphs that have  $n - 1$  edges, we have

$$\frac{S(n, e(G))e(G)!}{|\text{aut}G|} = \frac{\binom{n}{2}(n-1)!}{|\text{aut}G|}$$

But  $n!/|\text{aut}G|$  is the number of labelled graphs isomorphic to  $G$ . Therefore, summing over trees, we get

$$N(n, d) \geq \left( \frac{(n-1)n^{n-2}}{2} \right)^d.$$

The upper bound on  $N(n, d)$  is obtained by counting fully labelled pedigrees that do not even possibly admit a valid gender labelling.

We derive a lower bound on  $M(n, d)$  by enumerating a special subclass of general pedigrees that is described next. Consider pedigrees of depth  $d$  and order  $n$  that satisfy the conditions:

- (1) there are  $n$  vertices at each depth  $k \leq d$ ,
- (2) each vertex at depth  $k \leq d-2$  has exactly one parent at depth  $k+1$ ,
- (3) distinct vertices at depth  $k \leq d-2$  have distinct parents at depth  $k+1$ ,
- (4) at each depth  $k; k \leq d-1$ , there are  $n/2$  vertices of each gender,
- (5) the pedigree of depth 1 induced by vertices in  $X_{d-1} \cup X_d$  has no non-trivial automorphisms that fix vertices in  $X_{d-1}$ .

The conditions imply that given any vertex  $v$  at depth  $k; k \leq d-1$  there is a unique path of length  $k$  beginning at some vertex  $u$  in  $X_0$  and ending at  $v$ . Therefore, vertices at depth at most  $d-1$  are distinguishable. The last condition above makes the vertices at depth  $d$  distinguishable as well. Therefore, no two pedigrees described by the above conditions are isomorphic. This allows us to derive a lower bound on  $M(n, d)$ .

$$M(n, d) \geq \frac{(n-1)n^{n-2}}{2} \prod_{k=0}^{d-2} ((n/2)(d-1-k))^n,$$

where the first factor is a lower bound on the number of distinct pedigrees of depth 1 that are induced by  $X_{d-1} \cup X_d$ , vertices in  $X_{d-1}$  being labelled. For a vertex at depth  $k \leq d-2$ , the parent that is not at depth  $k+1$  may be chosen from the  $(n/2)(d-1-k)$  distinguishable vertices at depth  $k+2$  or more. This explains the second factor.

An upper bound on  $M(n, d)$  is obtained by counting the number of labelled directed graphs in which each vertex has out-degree 2.  $\square$

*Remark 4.* Steel and Hein give the information theoretic argument that if there are  $s$  segregating sites in DNA sequences obtained from  $n$  extant individuals, then there are  $4^{ns}$  possible combinations of sequences. Therefore,  $4^{ns}$  must be at least  $N(n, d)$  (or  $M(n, d)$ ) depending on what assumptions are made about pedigrees) to be able to reconstruct their pedigree up to depth  $d$ . They derive a lower bound on  $s$  given by

$(d/3) \log n$  for reconstruction of discrete generation constant population size pedigrees. They comment that in reality the number of sites required is likely to be much higher due to under-counting of isomorphism classes and due to the stochastic nature of sequence evolution. Theorem 2 gives an information theoretic lower bound on  $s$  that is about  $(d/2) \log n$  for discrete generation constant population size pedigrees, and a bound of about  $(d/2) \log(nd)$  for general pedigrees. Moreover, the bounds based on the upper bounds on  $N(n, d)$  and  $M(n, d)$  are only about  $d \log n$  and  $d \log(nd)$ , respectively, for discrete generation and general pedigrees.

*Remark 5.* If we assume that no vertex at depth  $k$  has a parent at depth more than  $k + t + 1$  then we have

$$M(n, d) \geq \frac{(n-1)n^{n-2}}{2} \prod_{k=0}^{d-t-1} (nt/2)^n \prod_{k=d-t}^{d-2} (n(d-k-1)/2)^n$$

This gives a lower bound of about  $(d/2) \log(nt)$  on the number of segregating sites required for pedigree reconstruction.

**Acknowledgements.** I would like to thank Mike Steel for many useful conversations, and for hosting me at the University of Canterbury. Oliver Will and Mike Steel read the manuscript carefully and made many helpful comments. I am supported by the Allan Wilson Centre for Molecular Ecology and Evolution, New Zealand.

## REFERENCES

- [1] L. Lovász. A note on the line reconstruction problem. *J. Combinatorial Theory Ser. B*, 13:309–310, 1972.
- [2] C. St. J. A. Nash-Williams. The reconstruction problem. In Lowell W. Beineke and Robin J. Wilson, editors, *Selected topics in graph theory*, pages 205–236. Academic Press Inc. [Harcourt Brace Jovanovich Publishers], London, 1978.
- [3] M. Steel and J. Hein. Reconstructing pedigrees: a combinatorial perspective. *Journal of Theoretical Biology*, 2006.

BIOMATHEMATICS RESEARCH CENTRE, MATHEMATICS AND COMPUTER SCIENCE BUILDING, UNIVERSITY OF CANTERBURY, PRIVATE BAG 4800, CHRISTCHURCH, NEW ZEALAND

*E-mail address:* bdthatte@gmail.com